SÃ©bastien KÃœry

List of Publications by Year
in descending order

114 papers	7,414 citations	87723 38 h-index	62479 80 g-index
130 all docs	130 docs citations	130 times ranked	13582 citing authors

#	Article	IF	Citations
1	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	1.1	9
2	Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder. American Journal of Human Genetics, 2022, 109, 361-372.	2.6	6
3	GestaltMatcher facilitates rare disease matching using facial phenotype descriptors. Nature Genetics, 2022, 54, 349-357.	9.4	73
4	Rare pathogenic variants in WNK3 cause X-linked intellectual disability. Genetics in Medicine, 2022, 24, 1941-1951.	1.1	5
5	SLITRK2 variants associated with neurodevelopmental disorders impair excitatory synaptic function and cognition in mice. Nature Communications, 2022, 13, .	5.8	6
6	NEXMIF encephalopathy: an X-linked disorder with male and female phenotypic patterns. Genetics in Medicine, 2021, 23, 363-373.	1.1	28
7	Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European Journal of Human Genetics, 2021, 29, 625-636.	1.4	17
8	Genetic architectures of proximal and distal colorectal cancer are partly distinct. Gut, 2021, 70, 1325-1334.	6.1	44
9	Disruption of RFX family transcription factors causes autism, attention-deficit/hyperactivity disorder, intellectual disability, and dysregulated behavior. Genetics in Medicine, 2021, 23, 1028-1040.	1.1	34
10	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	2.6	15
11	Missense variants in DPYSL5 cause a neurodevelopmental disorder with corpus callosum agenesis and cerebellar abnormalities. American Journal of Human Genetics, 2021, 108, 951-961.	2.6	26
12	Disruption of NEUROD2 causes a neurodevelopmental syndrome with autistic features via cell-autonomous defects in forebrain glutamatergic neurons. Molecular Psychiatry, 2021, 26, 6125-6148.	4.1	21
13	Neurodevelopmental Disorders (NDD) Caused by Genomic Alterations of the Ubiquitin-Proteasome System (UPS): the Possible Contribution of Immune Dysregulation to Disease Pathogenesis. Frontiers in Molecular Neuroscience, 2021, 14, 733012.	1.4	15
14	Partial Loss of USP9X Function Leads to a Male Neurodevelopmental and Behavioral Disorder Converging on Transforming Growth Factor Î² Signaling. Biological Psychiatry, 2020, 87, 100-112.	0.7	42
15	Cumulative Burden of Colorectal Cancerâ€“Associated Genetic Variants Is More Strongly Associated With Early-Onset vs Late-Onset Cancer. Gastroenterology, 2020, 158, 1274-1286.e12.	0.6	110
16	Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients. Science Advances, 2020, 6, .	4.7	43
17	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
18	Missense variant contribution to USP9X-female syndrome. Npj Genomic Medicine, 2020, 5, 53.	1.7	17

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19	De novo EIF2AK1 and EIF2AK2 Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. American Journal of Human Genetics, 2020, 106, 570-583.	2.6	37
20	Pathogenic DDX3X Mutations Impair RNA Metabolism and Neurogenesis during Fetal Cortical Development. Neuron, 2020, 106, 404-420.e8.	3.8	121
21	A dominant vimentin variant causes a rare syndrome with premature aging. European Journal of Human Genetics, 2020, 28, 1218-1230.	1.4	23
22	Novel Common Genetic Susceptibility Loci for Colorectal Cancer. Journal of the National Cancer Institute, 2019, 111, 146-157.	3.0	129
23	Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies. Genetics in Medicine, 2019, 21, 1797-1807.	1.1	41
24	Biallelic pathogenic variants in the lanosterol synthase gene LSS involved in the cholesterol biosynthesis cause alopecia with intellectual disability, a rare recessive neuroectodermal syndrome. Genetics in Medicine, 2019, 21, 2025-2035.	1.1	40
25	Enabling Global Clinical Collaborations on Identifiable Patient Data: The Minerva Initiative. Frontiers in Genetics, 2019, 10, 611.	1.1	14
26	Dermatological manifestations of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis (<scp>POIKTMP</scp>): a case series of 28 patients. British Journal of Dermatology, 2019, 181, 862-864.	1.4	8
27	Genetic variant predictors of gene expression provide new insight into risk of colorectal cancer. Human Genetics, 2019, 138, 307-326.	1.8	44
28	Missense Variants in the Histone Acetyltransferase Complex Component Gene TRRAP Cause Autism and Syndromic Intellectual Disability. American Journal of Human Genetics, 2019, 104, 530-541.	2.6	30
29	FAM111B Mutation Is Associated With Pancreatic Cancer Predisposition. Pancreas, 2019, 48, e41-e42.	0.5	20
30	Complex Compound Inheritance of Lethal Lung Developmental Disorders Due to Disruption of the TBX-FGF Pathway. American Journal of Human Genetics, 2019, 104, 213-228.	2.6	90
31	Discovery of common and rare genetic risk variants for colorectal cancer. Nature Genetics, 2019, 51, 76-87.	9.4	377
32	Clinical Presentation of a Complex Neurodevelopmental Disorder Caused by Mutations in ADNP. Biological Psychiatry, 2019, 85, 287-297.	0.7	108
33	Identification of shared genetic variants between schizophrenia and lung cancer. Scientific Reports, 2018, 8, 674.	1.6	33
34	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	2.6	51
35	Truncating Variants in NAA15 Are Associated with Variable Levels of Intellectual Disability, Autism Spectrum Disorder, and Congenital Anomalies. American Journal of Human Genetics, 2018, 102, 985-994.	2.6	59
36	<scp>ARL6IP1</scp> mutation causes congenital insensitivity to pain, acromutilation and spastic paraplegia. Clinical Genetics, 2018, 93, 169-172.	1.0	24

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37	Okurâ€Chung neurodevelopmental syndrome: Eight additional cases with implications on phenotype and genotype expansion. Clinical Genetics, 2018, 93, 880-890.	1.0	30
38	Transient neonatal zinc deficiency in exclusively breastfed preterm infants. Journal of Paediatrics and Child Health, 2018, 54, 319-322.	0.4	6
39	Cover Image, Volume 39, Issue 12. Human Mutation, 2018, 39, i-i.	1.1	0
40	The intellectual disability-associated CAMK2G p.Arg292Pro mutation acts as a pathogenic gain-of-function. Human Mutation, 2018, 39, 2008-2024.	1.1	25
41	Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome. American Journal of Human Genetics, 2018, 102, 1126-1142.	2.6	128
42	De Novo and Inherited Loss-of-Function Variants in TLK2: Clinical and Genotype-Phenotype Evaluation of a Distinct Neurodevelopmental Disorder. American Journal of Human Genetics, 2018, 102, 1195-1203.	2.6	37
43	Abstract 5268: Interactions between genetic predictors of gene expression and dietary factors associated with risk of colorectal cancer. , 2018, , .		0
44	Rare and low-frequency coding variants alter human adult height. Nature, 2017, 542, 186-190.	13.7	544
45	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	2.6	86
46	Biallelic Variants in OTUD6B Cause an Intellectual Disability Syndrome Associated with Seizures and Dysmorphic Features. American Journal of Human Genetics, 2017, 100, 676-688.	2.6	54
47	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	3.6	184
48	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	2.6	62
49	De Novo Missense Mutations in DHX30 Impair Global Translation and Cause a Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 101, 716-724.	2.6	66
50	De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability. American Journal of Human Genetics, 2017, 101, 768-788.	2.6	136
51	Characterization of zinc amino acid complexes for zinc delivery in vitro using Caco-2 cells and enterocytes from hiPSC. BioMetals, 2017, 30, 643-661.	1.8	60
52	Two novel variants in CNTNAP1 in two siblings presenting with congenital hypotonia and hypomyelinating neuropathy. European Journal of Human Genetics, 2017, 25, 150-152.	1.4	13
53	Fine-Mapping of Common Genetic Variants Associated with Colorectal Tumor Risk Identified Potential Functional Variants. PLoS ONE, 2016, 11, e0157521.	1.1	8
54	Identification of a Novel Mutation in the SLC39A4 Gene in a Case of Acrodermatitis Enteropathica. Acta Dermato-Venereologica, 2016, 96, 424-425.	0.6	4

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55	Clinical utility gene card for: acrodermatitis enteropathica â€“ update 2015. European Journal of Human Genetics, 2016, 24, 779-779.	1.4	7
56	Germline De Novo Mutations in GNB1 Cause Severe Neurodevelopmental Disability, Hypotonia, and Seizures. American Journal of Human Genetics, 2016, 98, 1001-1010.	2.6	102
57	Telomere structure and maintenance gene variants and risk of five cancer types. International Journal of Cancer, 2016, 139, 2655-2670.	2.3	43
58	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	2.6	45
59	CUGC for hereditary fibrosing poikiloderma with tendon contractures, myopathy, and pulmonary fibrosis (POIKTMP). European Journal of Human Genetics, 2016, 24, 779-779.	1.4	8
60	De Novo Truncating Mutations in the Kinetochore-Microtubules Attachment GeneCHAMP1Cause Syndromic Intellectual Disability. Human Mutation, 2016, 37, 354-358.	1.1	40
61	Clinical utility gene card for: Biotinidase deficiencyâ€”update 2015. European Journal of Human Genetics, 2016, 24, 3-5.	1.4	16
62	Identification of Susceptibility Loci and Genes for Colorectal Cancer Risk. Gastroenterology, 2016, 150, 1633-1645.	0.6	97
63	Genome-Wide Interaction Analyses between Genetic Variants and Alcohol Consumption and Smoking for Risk of Colorectal Cancer. PLoS Genetics, 2016, 12, e1006296.	1.5	38
64	PoÃ¯kilodermie hÃ©rÃ©ditaire fibrosante, myopathie rÃ©tractile et fibrose pulmonaire (POIKTMP). Les Cahiers De Myologie, 2016, , 12-14.	0.0	0
65	Expanding the clinical spectrum of hereditary fibrosing poikiloderma with tendon contractures, myopathy and pulmonary fibrosis due to FAM111B mutations. Orphanet Journal of Rare Diseases, 2015, 10, 135.	1.2	42
66	Risk factors for hepatocellular carcinoma in Caucasian patients with nonâ€viral cirrhosis: the importance of prior obesity. Liver International, 2015, 35, 1872-1876.	1.9	19
67	Mendelian randomization study of height and risk of colorectal cancer. International Journal of Epidemiology, 2015, 44, 662-672.	0.9	55
68	Autosomal-Recessive Intellectual Disability with Cerebellar Atrophy Syndrome Caused by Mutation of the Manganese and Zinc Transporter Gene SLC39A8. American Journal of Human Genetics, 2015, 97, 886-893.	2.6	171
69	Acute cytotoxicity of MIRA-1/NSC19630, a mutant p53-reactivating small molecule, against human normal and cancer cells via a caspase-9-dependent apoptosis. Cancer Letters, 2015, 359, 211-217.	3.2	34
70	Genome-wide association study of colorectal cancer identifies six new susceptibility loci. Nature Communications, 2015, 6, 7138.	5.8	138
71	Clinical Zinc Deficiency as Early Presentation of Wilson Disease. Journal of Pediatric Gastroenterology and Nutrition, 2015, 60, 457-459.	0.9	10
72	A genome-wide association study for colorectal cancer identifies a risk locus in 14q23.1. Human Genetics, 2015, 134, 1249-1262.	1.8	28

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73	Identification of a common variant with potential pleiotropic effect on risk of inflammatory bowel disease and colorectal cancer. Carcinogenesis, 2015, 36, 999-1007.	1.3	28
74	Acrodermatitis Enteropathica: A NovelSLC39A4Gene Mutation in a Patient with Normal Zinc Levels. Pediatric Dermatology, 2015, 32, e124-e125.	0.5	11
75	Pleiotropic effects of genetic risk variants for other cancers on colorectal cancer risk: PAGE, GECCO and CCFR consortia. Gut, 2014, 63, 800-807.	6.1	35
76	Trans-ethnic genome-wide association study of colorectal cancer identifies a new susceptibility locus in VTI1A. Nature Communications, 2014, 5, 4613.	5.8	72
77	Reappraisal of the soâ€called â€˜villous tumoursâ€™ of the rectosigmoid, based on histological, immunohistochemical and genotypic features. United European Gastroenterology Journal, 2014, 2, 307-314.	1.6	2
78	G.P.156. Neuromuscular Disorders, 2014, 24, 848.	0.3	0
79	Evaluation of the colorectal cancer risk conferred by rareUNC5Calleles. World Journal of Gastroenterology, 2014, 20, 204.	1.4	11
80	Abstract 2190: Fine-mapping of common genetic variants associated with colorectal tumor risk identified potential functional variants. , 2014, , .		0
81	Mutations in FAM111B Cause Hereditary Fibrosing Poikiloderma with Tendon Contracture, Myopathy, and Pulmonary Fibrosis. American Journal of Human Genetics, 2013, 93, 1100-1107.	2.6	76
82	751 Identification of a Common Variant With Potential Pleiotropic Effect on Risk of Inflammatory Bowel Disease and Colorectal Cancer. Gastroenterology, 2013, 144, S-136.	0.6	0
83	Genome-wide association analyses in east Asians identify new susceptibility loci for colorectal cancer. Nature Genetics, 2013, 45, 191-196.	9.4	173
84	Identification of Genetic Susceptibility Loci for Colorectal Tumors in a Genome-Wide Meta-analysis. Gastroenterology, 2013, 144, 799-807.e24.	0.6	292
85	Genetic Predictors of Circulating 25-Hydroxyvitamin D and Risk of Colorectal Cancer. Cancer Epidemiology Biomarkers and Prevention, 2013, 22, 2037-2046.	1.1	30
86	Clinical utility gene card for: acrodermatitis enteropathica. European Journal of Human Genetics, 2012, 20, 3-4.	1.4	8
87	Delineation of the infrequent mosaicism ofKRASmutational status in metastatic colorectal adenocarcinomas. Journal of Clinical Pathology, 2012, 65, 466-469.	1.0	14
88	Characterization of Geneâ€“Environment Interactions for Colorectal Cancer Susceptibility Loci. Cancer Research, 2012, 72, 2036-2044.	0.4	140
89	Clinical utility gene card for: Biotinidase deficiency. European Journal of Human Genetics, 2012, 20, 4-4.	1.4	6
90	PNPLA1 mutations cause autosomal recessive congenital ichthyosis in golden retriever dogs and humans. Nature Genetics, 2012, 44, 140-147.	9.4	208

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91	Meta-analysis of new genome-wide association studies of colorectal cancer risk. Human Genetics, 2012, 131, 217-234.	1.8	183
92	Genome-Wide Search for Gene-Gene Interactions in Colorectal Cancer. PLoS ONE, 2012, 7, e52535.	1.1	35
93	Ade novogermlineMLH1mutation in a Lynch syndrome patient with discordant immunohistochemical and molecular biology test results. World Journal of Gastroenterology, 2012, 18, 5635.	1.4	5
94	Acrodermatitis enteropathica: an uncommon differential diagnosis in childhood - first description of a new sequence variant. JDDG - Journal of the German Society of Dermatology, 2011, 9, 999-1002.	0.4	8
95	Acrodermatitis enteropathica: eine seltene dermatologische Differenzialdiagnose im Kindesalter - Erstbeschreibung zweier neuer Sequenzvarianten. JDDG - Journal of the German Society of Dermatology, 2011, 9, 999-1003.	0.4	13
96	Transient symptomatic zinc deficiency in a breast-fed infant: Relevance of a genetic study. Nutrition, 2011, 27, 1087-1089.	1.1	9
97	A Zinc Sulphate-Resistant Acrodermatitis Enteropathica Patient with a Novel Mutation in SLC39A4 Gene. JIMD Reports, 2011, 2, 25-28.	0.7	9
98	Colorectal Hamartomatous Polyposis and Ganglioneuromatosis in a Dog. Veterinary Pathology, 2011, 48, 1012-1015.	0.8	17
99	BAK1 gene variation and abdominal aortic aneurysms-results may have been prematurely overrated. Human Mutation, 2010, 31, 1174-1176.	1.1	6
100	Tropical medicine rounds: Acrodermatitis enteropathica: a review of 29 Tunisian cases. International Journal of Dermatology, 2010, 49, 1038-1044.	0.5	18
101	A large-scale meta-analysis to refine colorectal cancer risk estimates associated with MUTYH variants. British Journal of Cancer, 2010, 103, 1875-1884.	2.9	107
102	An update on mutations of theSLC39A4gene in acrodermatitis enteropathica. Human Mutation, 2009, 30, 926-933.	1.1	123
103	Three independent mutations in the TSC2 gene in a family with tuberous sclerosis. European Journal of Human Genetics, 2009, 17, 1165-1170.	1.4	8
104	Clinical, histopathological and genetic data of ichthyosis in the golden retriever: a prospective study. Journal of Small Animal Practice, 2009, 50, 227-235.	0.5	18
105	Low-penetrance alleles predisposing to sporadic colorectal cancers: a French case-controlled genetic association study. BMC Cancer, 2008, 8, 326.	1.1	82
106	The Thorough Screening of the MUTYH Gene in a Large French Cohort of Sporadic Colorectal Cancers. Genetic Testing and Molecular Biomarkers, 2007, 11, 373-380.	1.7	15
107	Combinations of Cytochrome P450 Gene Polymorphisms Enhancing the Risk for Sporadic Colorectal Cancer Related to Red Meat Consumption. Cancer Epidemiology Biomarkers and Prevention, 2007, 16, 1460-1467.	1.1	74
108	Genome-wide association scan identifies a colorectal cancer susceptibility locus on chromosome 8q24. Nature Genetics, 2007, 39, 989-994.	9.4	676

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109	A novel mutation of the SLC39A4 gene causing acrodermatitis enteropathica. British Journal of Dermatology, 2007, 157, 386-387.	1.4	8
110	Mutation analysis of the zinc transporter gene SLC30A4 reveals no association with periodic catatonia on chromosome 15q15. Journal of Neural Transmission, 2003, 110, 1329-1332.	1.4	9
111	Mutation spectrum of humanSLC39A4in a panel of patients with acrodermatitis enteropathica. Human Mutation, 2003, 22, 337-338.	1.1	70
112	A Novel Zinc-regulated Human Zinc Transporter, hZTL1, Is Localized to the Enterocyte Apical Membrane. Journal of Biological Chemistry, 2002, 277, 22789-22797.	1.6	123
113	Identification of SLC39A4, a gene involved in acrodermatitis enteropathica. Nature Genetics, 2002, 31, 239-240.	9.4	486
114	Expression pattern, genomic structure and evaluation of the human SLC30A4 gene as a candidate for acrodermatitis enteropathica. Human Genetics, 2001, 109, 178-185.	1.8	23